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PANEL HEREDITARY CANCER (CDH1, CTNNA1, MLH1, MSH2, MSH6, PMS2) DG-5.1.0 (7 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
CDH1	100%	100%	100%	100%	99.6%	Ovarian cancer, somatic, 167000;Blepharocheilodontic syndrome 1, 119580;Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215;Endometrial carcinoma, somatic, 608089;Breast cancer, lobular, somatic, 114480
CTNNA1	94.8%	94.8%	100%	100%	99.8%	Macular dystrophy, patterned, 2, 608970
EPCAM	100%	100%	100%	100%	99.3%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244
MLH1	100%	100%	100%	100%	99.7%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MSH2	90.6%	90.6%	100%	100%	99.6%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH6	100%	100%	100%	100%	99.5%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089

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PMS2	93.4%	93.4%	99.8%	99.6%	98.9%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors